# CISD2 gene

CDGSH iron sulfur domain 2

## Normal Function

The *CISD2* gene provides instructions for making a protein that is found in the outer membrane of cell structures called mitochondria. Mitochondria are involved in a wide variety of cellular activities, including energy production, chemical signaling, and regulation of cell growth and division. The exact function of the CISD2 protein is unknown, but it is thought to help keep mitochondria functioning normally.

## Health Conditions Related to Genetic Changes

### Wolfram syndrome

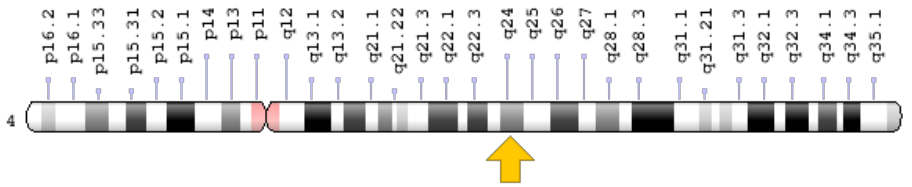
At least one mutation in the *CISD2* gene has been found to cause Wolfram syndrome. This condition is characterized by a lack of insulin leading to increased blood sugar (diabetes mellitus), a degeneration of nerves that carry information from the eyes to the brain (optic atrophy), and a number of other features involving the urinary tract, the brain, and hearing. People with this *CISD2* gene mutation also experience gastrointestinal ulcers and excessive bleeding after injury.

The *CISD2* gene mutation that causes Wolfram syndrome replaces the amino acid glutamic acid with the amino acid glutamine at position 37 in the CISD2 protein (written as Glu37Gln or E37Q). This mutation results in an abnormally small, nonfunctional CISD2 protein. As a result, the function of the mitochondria is impaired and they eventually break down. Since the mitochondria provide energy to cells, the loss of mitochondria leads to decreased energy for cells. Cells that do not have enough energy to function will eventually die. Cells with high energy demands, such as nerve cells in the brain, eyes, or gastrointestinal tract, are most susceptible to cell death due to reduced energy. The gradual loss of cells in various body systems likely causes the signs and symptoms of Wolfram syndrome. When Wolfram syndrome is caused by *CISD2* gene mutations, it is sometimes referred to as Wolfram syndrome type 2.

## Chromosomal Location

Cytogenetic Location: 4q24, which is the long (q) arm of chromosome 4 at position 24

Molecular Location: base pairs 102,868,978 to 102,892,807 on chromosome 4 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- CDGSH iron-sulfur domain-containing protein 2
- CISD2_HUMAN
- endoplasmic reticulum intermembrane small protein
- ERIS
- Miner1
- NAF-1
- nutrient-deprivation autophagy factor-1
- WFS2
- ZCD2
- zinc finger, CDGSH-type domain 2

## Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Mitochondria Are Bounded by a Double Membrane
  https://www.ncbi.nlm.nih.gov/books/NBK22511/#A2489
- Madame Curie Bioscience Database: Damage to Mitochondria
  https://www.ncbi.nlm.nih.gov/books/NBK6156/#A76012

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28CISD2%5BTIAB%5D%29+OR+%28%28ERIS%5BTIAB%5D%29+OR+%28WFS2%5BTIAB%5D%29+OR+%28ZCD2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CDGSH IRON SULFUR DOMAIN PROTEIN 2
  http://omim.org/entry/611507

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CISD2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CISD2%5Bgene%5D
- HGNC Gene Family: CDGSH iron sulfur domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/481
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=24212
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/493856
- UniProt
  http://www.uniprot.org/uniprot/Q8N5K1

## Sources for This Summary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  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/17846994
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2227919/
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  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2832857/


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